Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
DBT	Maple syrup urine disease	African/African American	99	1 in 404	1 in 40,000	1 in 160,000
		Latino/Admixed American	98	1 in 1,132	1 in 57,000	1 in 230,000
		Ashkenazi Jewish	97	1 in 1,640	1 in 55,000	1 in 220,000
		East Asian	98	1 in 1,277	1 in 64,000	1 in 260,000
		Finnish	99	1 in 1,099	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	99	1 in 294	1 in 29,000	1 in 120,000
		South Asian	99	1 in 1,090	1 in 110,000	1 in 440,000
		Other (population not assigned)	99	1 in 489	1 in 49,000	1 in 200,000
DCAF17	Neurodegeneration with brain iron accumulation disorder	African/African American	97	1 in 8,126	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	99	1 in 2,564	1 in 260,000	1 in 1,000,000
		Ashkenazi Jewish	99	1 in 199	1 in 20,000	1 in 80,000
		East Asian	99	1 in 1,757	1 in 180,000	1 in 720,000
		Finnish	99	1 in 1,800	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 1,053	1 in 110,000	1 in 440,000
		South Asian	99	1 in 2,479	1 in 250,000	1 in 1,000,000
		Other (population not assigned)	99	1 in 538	1 in 54,000	1 in 220,000
DCLRE1C	Omenn syndrome	African/African American	99	1 in 100	1 in 9,900	1 in 40,000
		Latino/Admixed American	98	1 in 973	1 in 49,000	1 in 200,000
		Ashkenazi Jewish	97	1 in 202	1 in 6,700	1 in 27,000
		East Asian	98	1 in 836	1 in 42,000	1 in 170,000
		Finnish	99	1 in 2,703	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 775	1 in 39,000	1 in 160,000
		South Asian	97	1 in 356	1 in 12,000	1 in 48,000
		Other (population not assigned)	97	1 in 203	1 in 6,700	1 in 27,000
DDB2	Xeroderma pigmentosum	African/African American	97	1 in 8,129	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	99	1 in 5,766	1 in 580,000	1 in 2,300,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 1,533	1 in 77,000	1 in 310,000
		Finnish	99	1 in 679	1 in 68,000	1 in 270,000
		Non-Finnish European/Caucasian	99	1 in 3,138	1 in 310,000	1 in 1,200,000
		South Asian	97	1 in 15,308	1 in 510,000	1 in 2,000,000
		Other (population not assigned)	97	1 in 3,071	1 in 100,000	1 in 400,000
DDC	Aromatic l-amino acid decarboxylase deficiency	African/African American	99	1 in 2,535	1 in 250,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 2,137	1 in 210,000	1 in 840,000
		Ashkenazi Jewish	99	1 in 279	1 in 28,000	1 in 110,000
		East Asian	97	1 in 1,839	1 in 61,000	1 in 240,000
		Finnish	99	1 in 1,361	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	99	1 in 976	1 in 98,000	1 in 390,000
		South Asian	99	1 in 1,180	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 1,527	1 in 150,000	1 in 600,000
DHCR7	Smith-Lemli-Opitz syndrome	African/African American	99	1 in 133	1 in 13,000	1 in 52,000
		Latino/Admixed American	99	1 in 96	1 in 9,500	1 in 38,000
		Ashkenazi Jewish	99	1 in 40	1 in 3,900	1 in 16,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		East Asian	99	1 in 273	1 in 27,000	1 in 110,000
		Finnish	99	1 in 203	1 in 20,000	1 in 80,000
		Non-Finnish European/Caucasian	99	1 in 47	1 in 4,600	1 in 18,000
		South Asian	99	1 in 470	1 in 47,000	1 in 190,000
		Other (population not assigned)	99	1 in 49	1 in 4,800	1 in 19,000
DHDDS	Retinitis pigmentosa	African/African American	99	1 in 200	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 4,291	1 in 430,000	1 in 1,700,000
		Ashkenazi Jewish	99	1 in 108	1 in 11,000	1 in 44,000
		East Asian	98	N/A	N/A	N/A
		Finnish	99	1 in 3,661	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	99	1 in 1,473	1 in 150,000	1 in 600,000
		South Asian	98	1 in 6,490	1 in 320,000	1 in 1,300,000
		Other (population not assigned)	99	1 in 945	1 in 94,000	1 in 380,000
DLAT	Pyruvate dehydrogenase deficiency	African/African American	99	1 in 1,244	1 in 120,000	1 in 480,000
		Latino/Admixed American	97	1 in 1,613	1 in 54,000	1 in 220,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 2,220	1 in 74,000	1 in 300,000
		Finnish	99	1 in 1,736	1 in 170,000	1 in 680,000
		Non-Finnish European/Caucasian	98	1 in 1,265	1 in 63,000	1 in 250,000
		South Asian	99	1 in 1,262	1 in 130,000	1 in 520,000
		Other (population not assigned)	99	1 in 602	1 in 60,000	1 in 240,000
DLD	Dihydrolipoamide dehydrogenase deficiency	African/African American	99	1 in 1,234	1 in 120,000	1 in 480,000
		Latino/Admixed American	99	1 in 1,692	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	99	1 in 61	1 in 6,000	1 in 24,000
		East Asian	98	1 in 2,239	1 in 110,000	1 in 440,000
		Finnish	99	1 in 1,796	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 968	1 in 97,000	1 in 390,000
		South Asian	98	1 in 1,293	1 in 65,000	1 in 260,000
		Other (population not assigned)	99	1 in 912	1 in 91,000	1 in 360,000
DMD	Dystrophinopathies, including Duchenne and Becker muscular dystrophy and X-linked cardiomyopathy	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
DNMT3B	Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome	African/African American	99	1 in 7,657	1 in 770,000	1 in 3,100,000
		Latino/Admixed American	99	1 in 3,433	1 in 340,000	1 in 1,400,000
		Ashkenazi Jewish	99	1 in 5,041	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 8,621	1 in 860,000	1 in 3,400,000
		Finnish	98	1 in 5,412	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 2,684	1 in 270,000	1 in 1,100,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		South Asian	99	1 in 1,175	1 in 120,000	1 in 480,000
		Other (population not assigned)	98	1 in 1,448	1 in 72,000	1 in 290,000
DOCK8	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 181	1 in 18,000	1 in 72,000
		Latino/Admixed American	99	1 in 339	1 in 34,000	1 in 140,000
		Ashkenazi Jewish	97	1 in 733	1 in 24,000	1 in 96,000
		East Asian	99	1 in 386	1 in 39,000	1 in 160,000
		Finnish	97	1 in 103	1 in 3,400	1 in 14,000
		Non-Finnish European/Caucasian	98	1 in 221	1 in 11,000	1 in 44,000
		South Asian	98	1 in 269	1 in 13,000	1 in 52,000
		Other (population not assigned)	98	1 in 164	1 in 8,200	1 in 33,000
DOK7	Congenital myasthenic syndrome	African/African American	98	1 in 255	1 in 13,000	1 in 52,000
		Latino/Admixed American	98	1 in 178	1 in 8,900	1 in 36,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 255	1 in 13,000	1 in 52,000
		Finnish	97	1 in 117	1 in 3,900	1 in 16,000
		Non-Finnish European/Caucasian	97	1 in 262	1 in 8,700	1 in 35,000
		South Asian	99	1 in 193	1 in 19,000	1 in 76,000
		Other (population not assigned)	98	1 in 356	1 in 18,000	1 in 72,000
DPYD	Dihydropyrimidine dehydrogenase deficiency	African/African American	99	1 in 970	1 in 97,000	1 in 390,000
		Latino/Admixed American	98	1 in 710	1 in 35,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 2,146	1 in 210,000	1 in 840,000
		East Asian	99	1 in 196	1 in 20,000	1 in 80,000
		Finnish	98	1 in 2,176	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 558	1 in 28,000	1 in 110,000
		South Asian	97	1 in 248	1 in 8,200	1 in 33,000
		Other (population not assigned)	98	1 in 1,532	1 in 77,000	1 in 310,000
DTNBP1	Hermansky-Pudlak syndrome	African/African American	98	1 in 379	1 in 19,000	1 in 76,000
		Latino/Admixed American	98	1 in 253	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	97	1 in 206	1 in 6,800	1 in 27,000
		East Asian	98	1 in 266	1 in 13,000	1 in 52,000
		Finnish	97	1 in 532	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	98	1 in 295	1 in 15,000	1 in 60,000
		South Asian	98	1 in 226	1 in 11,000	1 in 44,000
		Other (population not assigned)	97	1 in 228	1 in 7,600	1 in 30,000
DYSF	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	99	1 in 77	1 in 7,600	1 in 30,000
		Latino/Admixed American	99	1 in 81	1 in 8,000	1 in 32,000
		Ashkenazi Jewish	99	1 in 103	1 in 10,000	1 in 40,000
		East Asian	99	1 in 135	1 in 13,000	1 in 52,000
		Finnish	99	1 in 31	1 in 3,000	1 in 12,000
		Non-Finnish European/Caucasian	99	1 in 48	1 in 4,700	1 in 19,000
		South Asian	99	1 in 79	1 in 7,800	1 in 31,000
		Other (population not assigned)	99	1 in 65	1 in 6,400	1 in 26,000
EFEMP2	Cutis laxa	African/African American	99	1 in 261	1 in 26,000	1 in 100,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Latino/Admixed American	99	1 in 1,232	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 1,022	1 in 100,000	1 in 400,000
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,457	1 in 250,000	1 in 1,000,000
		South Asian	99	1 in 1,833	1 in 180,000	1 in 720,000
		Other (population not assigned)	99	1 in 1,529	1 in 150,000	1 in 600,000
EIF2B1	Leukoencephalopathy with vanishing white matter	African/African American	99	1 in 712	1 in 71,000	1 in 280,000
		Latino/Admixed American	99	1 in 524	1 in 52,000	1 in 210,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 919	1 in 92,000	1 in 370,000
		Finnish	99	1 in 704	1 in 70,000	1 in 280,000
		Non-Finnish European/Caucasian	99	1 in 153	1 in 15,000	1 in 60,000
		South Asian	99	1 in 283	1 in 28,000	1 in 110,000
		Other (population not assigned)	99	1 in 337	1 in 34,000	1 in 140,000
EIF2B2	Leukoencephalopathy with vanishing white matter	African/African American	99	1 in 259	1 in 26,000	1 in 100,000
		Latino/Admixed American	99	1 in 482	1 in 48,000	1 in 190,000
		Ashkenazi Jewish	98	1 in 993	1 in 50,000	1 in 200,000
		East Asian	99	1 in 212	1 in 21,000	1 in 84,000
		Finnish	99	1 in 113	1 in 11,000	1 in 44,000
		Non-Finnish European/Caucasian	99	1 in 227	1 in 23,000	1 in 92,000
		South Asian	99	1 in 543	1 in 54,000	1 in 220,000
		Other (population not assigned)	99	1 in 221	1 in 22,000	1 in 88,000
EIF2B3	Leukoencephalopathy with vanishing white matter	African/African American	97	1 in 527	1 in 18,000	1 in 72,000
		Latino/Admixed American	97	1 in 147	1 in 4,900	1 in 20,000
		Ashkenazi Jewish	97	1 in 161	1 in 5,300	1 in 21,000
		East Asian	97	1 in 186	1 in 6,200	1 in 25,000
		Finnish	97	1 in 932	1 in 31,000	1 in 120,000
		Non-Finnish European/Caucasian	97	1 in 414	1 in 14,000	1 in 56,000
		South Asian	97	1 in 306	1 in 10,000	1 in 40,000
		Other (population not assigned)	97	1 in 184	1 in 6,100	1 in 24,000
EIF2B4	Leukoencephalopathy with vanishing white matter	African/African American	98	1 in 2,323	1 in 120,000	1 in 480,000
		Latino/Admixed American	99	1 in 672	1 in 67,000	1 in 270,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 569	1 in 57,000	1 in 230,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,121	1 in 210,000	1 in 840,000
		South Asian	98	1 in 3,062	1 in 150,000	1 in 600,000
		Other (population not assigned)	99	1 in 2,744	1 in 270,000	1 in 1,100,000
EIF2B5	Leukoencephalopathy with vanishing white matter	African/African American	98	1 in 50	1 in 2,500	1 in 10,000
		Latino/Admixed American	98	1 in 61	1 in 3,000	1 in 12,000
		Ashkenazi Jewish	99	1 in 107	1 in 11,000	1 in 44,000
		East Asian	99	1 in 105	1 in 10,000	1 in 40,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		Finnish	99	1 in 50	1 in 4,900	1 in 20,000
		Non-Finnish European/Caucasian	98	1 in 63	1 in 3,100	1 in 12,000
		South Asian	98	1 in 82	1 in 4,100	1 in 16,000
		Other (population not assigned)	98	1 in 67	1 in 3,300	1 in 13,000
ELP1	Familial dysautonomia	African/African American	99	1 in 594	1 in 59,000	1 in 240,000
		Latino/Admixed American	98	1 in 808	1 in 40,000	1 in 160,000
		Ashkenazi Jewish	99	1 in 37	1 in 3,600	1 in 14,000
		East Asian	99	1 in 657	1 in 66,000	1 in 260,000
		Finnish	98	1 in 3,586	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	99	1 in 782	1 in 78,000	1 in 310,000
		South Asian	99	1 in 853	1 in 85,000	1 in 340,000
		Other (population not assigned)	98	1 in 410	1 in 20,000	1 in 80,000
EMD	Emery-Dreifuss muscular dystrophy	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
ERCC2	Xeroderma pigmentosum	African/African American	99	1 in 222	1 in 22,000	1 in 88,000
		Latino/Admixed American	98	1 in 243	1 in 12,000	1 in 48,000
		Ashkenazi Jewish	99	1 in 95	1 in 9,400	1 in 38,000
		East Asian	97	1 in 49	1 in 1,600	1 in 6,400
		Finnish	99	1 in 490	1 in 49,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 154	1 in 15,000	1 in 60,000
		South Asian	99	1 in 54	1 in 5,300	1 in 21,000
		Other (population not assigned)	98	1 in 117	1 in 5,800	1 in 23,000
ERCC3	Xeroderma pigmentosum	African/African American	99	1 in 533	1 in 53,000	1 in 210,000
		Latino/Admixed American	98	1 in 363	1 in 18,000	1 in 72,000
		Ashkenazi Jewish	99	1 in 55	1 in 5,400	1 in 22,000
		East Asian	97	1 in 486	1 in 16,000	1 in 64,000
		Finnish	98	1 in 2,164	1 in 110,000	1 in 440,000
		Non-Finnish European/Caucasian	98	1 in 436	1 in 22,000	1 in 88,000
		South Asian	98	1 in 958	1 in 48,000	1 in 190,000
		Other (population not assigned)	98	1 in 362	1 in 18,000	1 in 72,000
ERCC4	Xeroderma pigmentosum	African/African American	99	1 in 672	1 in 67,000	1 in 270,000
		Latino/Admixed American	98	1 in 1,066	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	99	1 in 560	1 in 56,000	1 in 220,000
		East Asian	99	1 in 572	1 in 57,000	1 in 230,000
		Finnish	98	1 in 899	1 in 45,000	1 in 180,000
		Non-Finnish European/Caucasian	99	1 in 313	1 in 31,000	1 in 120,000
		South Asian	98	1 in 567	1 in 28,000	1 in 110,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		Other (population not assigned)	99	1 in 276	1 in 28,000	1 in 110,000
ERCC5	Xeroderma pigmentosum	African/African American	98	1 in 878	1 in 44,000	1 in 180,000
		Latino/Admixed American	98	1 in 909	1 in 45,000	1 in 180,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 703	1 in 70,000	1 in 280,000
		Finnish	99	1 in 10,385	1 in 1,000,000	1 in 4,000,000
		Non-Finnish European/Caucasian	98	1 in 970	1 in 48,000	1 in 190,000
		South Asian	99	1 in 279	1 in 28,000	1 in 110,000
		Other (population not assigned)	99	1 in 3,065	1 in 310,000	1 in 1,200,000
ERCC6	Cockayne syndrome	African/African American	98	1 in 419	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 214	1 in 21,000	1 in 84,000
		Ashkenazi Jewish	99	1 in 250	1 in 25,000	1 in 100,000
		East Asian	98	1 in 241	1 in 12,000	1 in 48,000
		Finnish	99	1 in 360	1 in 36,000	1 in 140,000
		Non-Finnish European/Caucasian	99	1 in 335	1 in 33,000	1 in 130,000
		South Asian	99	1 in 431	1 in 43,000	1 in 170,000
		Other (population not assigned)	99	1 in 431	1 in 43,000	1 in 170,000
ERCC8	Cockayne syndrome	African/African American	99	1 in 218	1 in 22,000	1 in 88,000
		Latino/Admixed American	99	1 in 1,557	1 in 160,000	1 in 640,000
		Ashkenazi Jewish	99	1 in 235	1 in 23,000	1 in 92,000
		East Asian	99	1 in 1,006	1 in 100,000	1 in 400,000
		Finnish	99	1 in 3,607	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 1,080	1 in 110,000	1 in 440,000
		South Asian	98	1 in 1,387	1 in 69,000	1 in 280,000
		Other (population not assigned)	99	1 in 3,038	1 in 300,000	1 in 1,200,000
ETFA	Glutaric acidemia type II	African/African American	97	1 in 172	1 in 5,700	1 in 23,000
		Latino/Admixed American	99	1 in 1,149	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	99	1 in 2,445	1 in 240,000	1 in 960,000
		East Asian	99	1 in 561	1 in 56,000	1 in 220,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 733	1 in 73,000	1 in 290,000
		South Asian	99	1 in 911	1 in 91,000	1 in 360,000
		Other (population not assigned)	98	1 in 837	1 in 42,000	1 in 170,000
ETFB	Glutaric acidemia type II	African/African American	99	1 in 8,122	1 in 810,000	1 in 3,200,000
		Latino/Admixed American	98	1 in 821	1 in 41,000	1 in 160,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 9,195	1 in 920,000	1 in 3,700,000
		Finnish	97	1 in 5,406	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 1,593	1 in 80,000	1 in 320,000
		South Asian	98	1 in 1,021	1 in 51,000	1 in 200,000
		Other (population not assigned)	98	1 in 1,448	1 in 72,000	1 in 290,000
ETFDH	Glutaric acidemia type II	African/African American	99	1 in 374	1 in 37,000	1 in 150,000
		Latino/Admixed American	98	1 in 312	1 in 16,000	1 in 64,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Ashkenazi Jewish	99	1 in 406	1 in 41,000	1 in 160,000
		East Asian	99	1 in 235	1 in 23,000	1 in 92,000
		Finnish	99	1 in 531	1 in 53,000	1 in 210,000
		Non-Finnish European/Caucasian	99	1 in 389	1 in 39,000	1 in 160,000
		South Asian	99	1 in 1,075	1 in 110,000	1 in 440,000
		Other (population not assigned)	99	1 in 407	1 in 41,000	1 in 160,000
ETHE1	Ethylmalonic encephalopathy	African/African American	99	1 in 833	1 in 83,000	1 in 330,000
		Latino/Admixed American	99	1 in 521	1 in 52,000	1 in 210,000
		Ashkenazi Jewish	99	1 in 4,985	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 1,417	1 in 140,000	1 in 560,000
		Finnish	99	1 in 2,006	1 in 200,000	1 in 800,000
		Non-Finnish European/Caucasian	99	1 in 953	1 in 95,000	1 in 380,000
		South Asian	99	1 in 782	1 in 78,000	1 in 310,000
		Other (population not assigned)	99	1 in 505	1 in 50,000	1 in 200,000
EXOSC3	Pontocerebellar hypoplasia	African/African American	98	1 in 507	1 in 25,000	1 in 100,000
		Latino/Admixed American	99	1 in 287	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	98	1 in 719	1 in 36,000	1 in 140,000
		East Asian	97	1 in 336	1 in 11,000	1 in 44,000
		Finnish	99	1 in 43	1 in 4,200	1 in 17,000
		Non-Finnish European/Caucasian	99	1 in 139	1 in 14,000	1 in 56,000
		South Asian	99	1 in 213	1 in 21,000	1 in 84,000
		Other (population not assigned)	99	1 in 149	1 in 15,000	1 in 60,000
EYS	Retinitis pigmentosa	African/African American	99	1 in 35	1 in 3,400	1 in 14,000
		Latino/Admixed American	99	1 in 73	1 in 7,200	1 in 29,000
		Ashkenazi Jewish	99	1 in 38	1 in 3,700	1 in 15,000
		East Asian	99	1 in 48	1 in 4,700	1 in 19,000
		Finnish	98	1 in 37	1 in 1,800	1 in 7,200
		Non-Finnish European/Caucasian	99	1 in 97	1 in 9,600	1 in 38,000
		South Asian	99	1 in 175	1 in 17,000	1 in 68,000
		Other (population not assigned)	99	1 in 48	1 in 4,700	1 in 19,000
FA2H	Neurodegeneration with brain iron accumulation disorder	African/African American	97	1 in 455	1 in 15,000	1 in 60,000
		Latino/Admixed American	98	1 in 1,143	1 in 57,000	1 in 230,000
		Ashkenazi Jewish	97	1 in 3,630	1 in 120,000	1 in 480,000
		East Asian	98	1 in 4,124	1 in 210,000	1 in 840,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,669	1 in 170,000	1 in 680,000
		South Asian	98	1 in 2,423	1 in 120,000	1 in 480,000
		Other (population not assigned)	99	1 in 1,448	1 in 140,000	1 in 560,000
FAH	Tyrosinemia type I	African/African American	99	1 in 765	1 in 76,000	1 in 300,000
		Latino/Admixed American	99	1 in 680	1 in 68,000	1 in 270,000
		Ashkenazi Jewish	99	1 in 130	1 in 13,000	1 in 52,000
		East Asian	99	1 in 8,621	1 in 860,000	1 in 3,400,000
		Finnish	99	1 in 295	1 in 29,000	1 in 120,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Non-Finnish European/Caucasian	99	1 in 319	1 in 32,000	1 in 130,000
		South Asian	99	1 in 549	1 in 55,000	1 in 220,000
		Other (population not assigned)	99	1 in 347	1 in 35,000	1 in 140,000
FAM126A	Hypomyelination and congenital cataract	African/African American	97	1 in 1,999	1 in 67,000	1 in 270,000
		Latino/Admixed American	98	1 in 2,458	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	1 in 1,402	1 in 140,000	1 in 560,000
		East Asian	99	1 in 4,595	1 in 460,000	1 in 1,800,000
		Finnish	97	1 in 8,219	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 874	1 in 44,000	1 in 180,000
		South Asian	97	1 in 717	1 in 24,000	1 in 96,000
		Other (population not assigned)	98	1 in 780	1 in 39,000	1 in 160,000
FAM161A	Retinitis pigmentosa	African/African American	97	1 in 1,104	1 in 37,000	1 in 150,000
		Latino/Admixed American	98	1 in 488	1 in 24,000	1 in 96,000
		Ashkenazi Jewish	97	1 in 235	1 in 7,800	1 in 31,000
		East Asian	99	1 in 1,450	1 in 140,000	1 in 560,000
		Finnish	97	1 in 558	1 in 19,000	1 in 76,000
		Non-Finnish European/Caucasian	99	1 in 402	1 in 40,000	1 in 160,000
		South Asian	98	1 in 716	1 in 36,000	1 in 140,000
		Other (population not assigned)	97	1 in 230	1 in 7,600	1 in 30,000
FANCA	Fanconi anemia	African/African American	99	1 in 88	1 in 8,700	1 in 35,000
		Latino/Admixed American	99	1 in 154	1 in 15,000	1 in 60,000
		Ashkenazi Jewish	99	1 in 168	1 in 17,000	1 in 68,000
		East Asian	98	1 in 87	1 in 4,300	1 in 17,000
		Finnish	99	1 in 228	1 in 23,000	1 in 92,000
		Non-Finnish European/Caucasian	99	1 in 108	1 in 11,000	1 in 44,000
		South Asian	99	1 in 104	1 in 10,000	1 in 40,000
		Other (population not assigned)	99	1 in 153	1 in 15,000	1 in 60,000
FANCB	Fanconi anemia	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
FANCC	Fanconi anemia	African/African American	98	1 in 659	1 in 33,000	1 in 130,000
		Latino/Admixed American	98	1 in 1,197	1 in 60,000	1 in 240,000
		Ashkenazi Jewish	99	1 in 84	1 in 8,300	1 in 33,000
		East Asian	99	1 in 2,995	1 in 300,000	1 in 1,200,000
		Finnish	98	1 in 5,493	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	98	1 in 548	1 in 27,000	1 in 110,000
		South Asian	99	1 in 960	1 in 96,000	1 in 380,000
		Other (population not assigned)	99	1 in 1,368	1 in 140,000	1 in 560,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
FANCD2	Fanconi anemia	African/African American	98	1 in 109	1 in 5,400	1 in 22,000
		Latino/Admixed American	99	1 in 322	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	98	1 in 414	1 in 21,000	1 in 84,000
		East Asian	98	1 in 372	1 in 19,000	1 in 76,000
		Finnish	98	1 in 256	1 in 13,000	1 in 52,000
		Non-Finnish European/Caucasian	98	1 in 177	1 in 8,800	1 in 35,000
		South Asian	98	1 in 437	1 in 22,000	1 in 88,000
		Other (population not assigned)	98	1 in 112	1 in 5,600	1 in 22,000
FANCE	Fanconi anemia	African/African American	99	1 in 427	1 in 43,000	1 in 170,000
		Latino/Admixed American	98	1 in 1,434	1 in 72,000	1 in 290,000
		Ashkenazi Jewish	99	1 in 5,030	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 1,148	1 in 110,000	1 in 440,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 916	1 in 92,000	1 in 370,000
		South Asian	98	1 in 874	1 in 44,000	1 in 180,000
		Other (population not assigned)	97	1 in 984	1 in 33,000	1 in 130,000
FANCF	Fanconi anemia	African/African American	98	1 in 1,104	1 in 55,000	1 in 220,000
		Latino/Admixed American	99	1 in 1,909	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 1,836	1 in 92,000	1 in 370,000
		Finnish	97	1 in 10,825	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	98	1 in 909	1 in 45,000	1 in 180,000
		South Asian	98	1 in 1,707	1 in 85,000	1 in 340,000
		Other (population not assigned)	99	1 in 1,022	1 in 100,000	1 in 400,000
FANCG	Fanconi anemia	African/African American	97	1 in 605	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 1,550	1 in 150,000	1 in 600,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,087	1 in 110,000	1 in 440,000
		Finnish	97	1 in 1,423	1 in 47,000	1 in 190,000
		Non-Finnish European/Caucasian	98	1 in 697	1 in 35,000	1 in 140,000
		South Asian	98	1 in 1,166	1 in 58,000	1 in 230,000
		Other (population not assigned)	97	1 in 930	1 in 31,000	1 in 120,000
FANCI	Fanconi anemia	African/African American	98	1 in 404	1 in 20,000	1 in 80,000
		Latino/Admixed American	98	1 in 524	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 5,038	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 459	1 in 46,000	1 in 180,000
		Finnish	98	1 in 113	1 in 5,600	1 in 22,000
		Non-Finnish European/Caucasian	99	1 in 557	1 in 56,000	1 in 220,000
		South Asian	98	1 in 373	1 in 19,000	1 in 76,000
		Other (population not assigned)	98	1 in 511	1 in 26,000	1 in 100,000
FANCL	Fanconi anemia	African/African American	97	1 in 295	1 in 9,800	1 in 39,000
		Latino/Admixed American	97	1 in 79	1 in 2,600	1 in 10,000
		Ashkenazi Jewish	97	1 in 68	1 in 2,200	1 in 8,800

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		East Asian	98	1 in 1,530	1 in 76,000	1 in 300,000
		Finnish	97	1 in 263	1 in 8,700	1 in 35,000
		Non-Finnish European/Caucasian	97	1 in 102	1 in 3,400	1 in 14,000
		South Asian	97	1 in 288	1 in 9,600	1 in 38,000
		Other (population not assigned)	97	1 in 82	1 in 2,700	1 in 11,000
FBXL4	Leigh syndrome, autosomal recessive	African/African American	99	1 in 697	1 in 70,000	1 in 280,000
		Latino/Admixed American	98	1 in 727	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	97	1 in 1,642	1 in 55,000	1 in 220,000
		East Asian	98	1 in 1,278	1 in 64,000	1 in 260,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 913	1 in 91,000	1 in 360,000
		South Asian	99	1 in 2,180	1 in 220,000	1 in 880,000
		Other (population not assigned)	99	1 in 2,734	1 in 270,000	1 in 1,100,000
FHL1	Emery-Dreifuss muscular dystrophy	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
FKBP10	Osteogenesis imperfecta, autosomal recessive	African/African American	97	1 in 641	1 in 21,000	1 in 84,000
		Latino/Admixed American	99	1 in 624	1 in 62,000	1 in 250,000
		Ashkenazi Jewish	99	1 in 839	1 in 84,000	1 in 340,000
		East Asian	99	1 in 457	1 in 46,000	1 in 180,000
		Finnish	97	1 in 1,591	1 in 53,000	1 in 210,000
		Non-Finnish European/Caucasian	98	1 in 1,025	1 in 51,000	1 in 200,000
		South Asian	98	1 in 803	1 in 40,000	1 in 160,000
		Other (population not assigned)	98	1 in 868	1 in 43,000	1 in 170,000
FKRP	Limb-girdle muscular dystrophy, autosomal recessive	African/African American	99	1 in 3,032	1 in 300,000	1 in 1,200,000
		Latino/Admixed American	99	1 in 424	1 in 42,000	1 in 170,000
		Ashkenazi Jewish	99	1 in 4,031	1 in 400,000	1 in 1,600,000
		East Asian	99	1 in 248	1 in 25,000	1 in 100,000
		Finnish	99	1 in 318	1 in 32,000	1 in 130,000
		Non-Finnish European/Caucasian	99	1 in 193	1 in 19,000	1 in 76,000
		South Asian	99	1 in 1,429	1 in 140,000	1 in 560,000
		Other (population not assigned)	99	1 in 231	1 in 23,000	1 in 92,000
FKTN	Walker-Warburg syndrome and other FKTN related dystrophies	African/African American	99	1 in 802	1 in 80,000	1 in 320,000
		Latino/Admixed American	97	1 in 345	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	97	1 in 62	1 in 2,000	1 in 8,000
		East Asian	99	1 in 766	1 in 77,000	1 in 310,000
		Finnish	99	1 in 127	1 in 13,000	1 in 52,000
		Non-Finnish European/Caucasian	99	1 in 931	1 in 93,000	1 in 370,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		South Asian	99	1 in 407	1 in 41,000	1 in 160,000
		Other (population not assigned)	98	1 in 480	1 in 24,000	1 in 96,000
FMR1	Fragile X syndrome	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
FOXN1	Severe combined immunodeficiency (SCID)	African/African American	97	1 in 1,161	1 in 39,000	1 in 160,000
		Latino/Admixed American	99	1 in 5,702	1 in 570,000	1 in 2,300,000
		Ashkenazi Jewish	97	1 in 336	1 in 11,000	1 in 44,000
		East Asian	98	1 in 4,565	1 in 230,000	1 in 920,000
		Finnish	97	1 in 5,411	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	98	1 in 4,349	1 in 220,000	1 in 880,000
		South Asian	97	1 in 7,652	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	97	1 in 3,063	1 in 100,000	1 in 400,000
FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy	African/African American	98	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	N/A	N/A	N/A
		South Asian	98	N/A	N/A	N/A
		Other (population not assigned)	98	N/A	N/A	N/A
FOXRED1	Leigh syndrome, autosomal recessive	African/African American	99	1 in 2,646	1 in 260,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 482	1 in 48,000	1 in 190,000
		Ashkenazi Jewish	97	1 in 549	1 in 18,000	1 in 72,000
		East Asian	99	1 in 180	1 in 18,000	1 in 72,000
		Finnish	99	1 in 1,326	1 in 130,000	1 in 520,000
		Non-Finnish European/Caucasian	98	1 in 439	1 in 22,000	1 in 88,000
		South Asian	99	1 in 145	1 in 14,000	1 in 56,000
		Other (population not assigned)	98	1 in 552	1 in 28,000	1 in 110,000
FRAS1	Fraser syndrome	African/African American	98	1 in 259	1 in 13,000	1 in 52,000
		Latino/Admixed American	97	1 in 111	1 in 3,700	1 in 15,000
		Ashkenazi Jewish	98	1 in 492	1 in 25,000	1 in 100,000
		East Asian	98	1 in 156	1 in 7,800	1 in 31,000
		Finnish	98	1 in 898	1 in 45,000	1 in 180,000
		Non-Finnish European/Caucasian	98	1 in 300	1 in 15,000	1 in 60,000
		South Asian	98	1 in 174	1 in 8,700	1 in 35,000
		Other (population not assigned)	97	1 in 228	1 in 7,600	1 in 30,000
FREM2	Fraser syndrome	African/African American	99	1 in 677	1 in 68,000	1 in 270,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Latino/Admixed American	99	1 in 279	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	99	1 in 315	1 in 31,000	1 in 120,000
		East Asian	99	1 in 518	1 in 52,000	1 in 210,000
		Finnish	99	1 in 89	1 in 8,800	1 in 35,000
		Non-Finnish European/Caucasian	99	1 in 115	1 in 11,000	1 in 44,000
		South Asian	99	1 in 49	1 in 4,800	1 in 19,000
		Other (population not assigned)	99	1 in 134	1 in 13,000	1 in 52,000
FUCA1	Fucosidosis	African/African American	98	1 in 2,032	1 in 100,000	1 in 400,000
		Latino/Admixed American	99	1 in 1,233	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	99	1 in 5,034	1 in 500,000	1 in 2,000,000
		East Asian	99	1 in 2,875	1 in 290,000	1 in 1,200,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 1,149	1 in 110,000	1 in 440,000
		South Asian	99	1 in 2,554	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	99	1 in 2,056	1 in 210,000	1 in 840,000
G6PC	Glycogen storage disease type I	African/African American	99	1 in 985	1 in 98,000	1 in 390,000
		Latino/Admixed American	98	1 in 374	1 in 19,000	1 in 76,000
		Ashkenazi Jewish	99	1 in 74	1 in 7,300	1 in 29,000
		East Asian	99	1 in 212	1 in 21,000	1 in 84,000
		Finnish	99	1 in 529	1 in 53,000	1 in 210,000
		Non-Finnish European/Caucasian	99	1 in 276	1 in 28,000	1 in 110,000
		South Asian	99	1 in 15,385	1 in 1,500,000	1 in 6,000,000
		Other (population not assigned)	99	1 in 474	1 in 47,000	1 in 190,000
GAA	Pompe disease	African/African American	99	1 in 104	1 in 10,000	1 in 40,000
		Latino/Admixed American	99	1 in 111	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	99	1 in 82	1 in 8,100	1 in 32,000
		East Asian	99	1 in 51	1 in 5,000	1 in 20,000
		Finnish	99	1 in 201	1 in 20,000	1 in 80,000
		Non-Finnish European/Caucasian	99	1 in 63	1 in 6,200	1 in 25,000
		South Asian	99	1 in 146	1 in 15,000	1 in 60,000
		Other (population not assigned)	99	1 in 104	1 in 10,000	1 in 40,000
GALC	Krabbe disease	African/African American	99	1 in 164	1 in 16,000	1 in 64,000
		Latino/Admixed American	99	1 in 200	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	99	1 in 533	1 in 53,000	1 in 210,000
		East Asian	99	1 in 46	1 in 4,500	1 in 18,000
		Finnish	99	1 in 152	1 in 15,000	1 in 60,000
		Non-Finnish European/Caucasian	99	1 in 80	1 in 7,900	1 in 32,000
		South Asian	99	1 in 35	1 in 3,400	1 in 14,000
		Other (population not assigned)	99	1 in 88	1 in 8,700	1 in 35,000
GALE	Galactosemia	African/African American	99	1 in 2,624	1 in 260,000	1 in 1,000,000
		Latino/Admixed American	99	1 in 1,217	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	97	1 in 4,888	1 in 160,000	1 in 640,000
		East Asian	99	1 in 369	1 in 37,000	1 in 150,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Finnish	97	1 in 10,825	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 2,011	1 in 200,000	1 in 800,000
		South Asian	99	1 in 2,541	1 in 250,000	1 in 1,000,000
		Other (population not assigned)	99	1 in 915	1 in 91,000	1 in 360,000
GALK1	Galactosemia	African/African American	99	1 in 652	1 in 65,000	1 in 260,000
		Latino/Admixed American	97	1 in 57	1 in 1,900	1 in 7,600
		Ashkenazi Jewish	99	1 in 453	1 in 45,000	1 in 180,000
		East Asian	99	1 in 430	1 in 43,000	1 in 170,000
		Finnish	99	1 in 2,150	1 in 210,000	1 in 840,000
		Non-Finnish European/Caucasian	98	1 in 1,165	1 in 58,000	1 in 230,000
		South Asian	97	1 in 520	1 in 17,000	1 in 68,000
		Other (population not assigned)	98	1 in 1,523	1 in 76,000	1 in 300,000
GALNS	Mucopolysaccharidosis type IVA	African/African American	99	1 in 548	1 in 55,000	1 in 220,000
		Latino/Admixed American	99	1 in 317	1 in 32,000	1 in 130,000
		Ashkenazi Jewish	99	1 in 824	1 in 82,000	1 in 330,000
		East Asian	99	1 in 263	1 in 26,000	1 in 100,000
		Finnish	98	1 in 277	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 311	1 in 31,000	1 in 120,000
		South Asian	98	1 in 317	1 in 16,000	1 in 64,000
		Other (population not assigned)	99	1 in 237	1 in 24,000	1 in 96,000
GALT	Galactosemia	African/African American	99	1 in 104	1 in 10,000	1 in 40,000
		Latino/Admixed American	99	1 in 142	1 in 14,000	1 in 56,000
		Ashkenazi Jewish	99	1 in 2,489	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 687	1 in 69,000	1 in 280,000
		Finnish	99	1 in 5,575	1 in 560,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 132	1 in 13,000	1 in 52,000
		South Asian	99	1 in 385	1 in 38,000	1 in 150,000
		Other (population not assigned)	99	1 in 235	1 in 23,000	1 in 92,000
GAMT	Cerebral creatine deficiency syndromes	African/African American	98	1 in 897	1 in 45,000	1 in 180,000
		Latino/Admixed American	98	1 in 3,195	1 in 160,000	1 in 640,000
		Ashkenazi Jewish	97	1 in 1,395	1 in 46,000	1 in 180,000
		East Asian	97	1 in 1,356	1 in 45,000	1 in 180,000
		Finnish	97	1 in 9,400	1 in 310,000	1 in 1,200,000
		Non-Finnish European/Caucasian	99	1 in 417	1 in 42,000	1 in 170,000
		South Asian	99	1 in 2,581	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	98	1 in 547	1 in 27,000	1 in 110,000
GATM	Cerebral creatine deficiency syndromes	African/African American	98	1 in 4,059	1 in 200,000	1 in 800,000
		Latino/Admixed American	98	N/A	N/A	N/A
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 2,225	1 in 220,000	1 in 880,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 6,747	1 in 670,000	1 in 2,700,000
		South Asian	99	1 in 15,299	1 in 1,500,000	1 in 6,000,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Other (population not assigned)	98	N/A	N/A	N/A
GBA	Gaucher disease	African/African American	87	1 in 139	1 in 1,100	1 in 4,400
		Latino/Admixed American	87	1 in 148	1 in 1,100	1 in 4,400
		Ashkenazi Jewish	97	1 in 15	1 in 470	1 in 1,900
		East Asian	87	1 in 166	1 in 1,300	1 in 5,200
		Finnish	87	1 in 191	1 in 1,500	1 in 6,000
		Non-Finnish European/Caucasian	87	1 in 138	1 in 1,100	1 in 4,400
		South Asian	87	1 in 202	1 in 1,500	1 in 6,000
		Other (population not assigned)	87	1 in 163	1 in 1,200	1 in 4,800
GBE1	Glycogen storage disease type IV	African/African American	98	1 in 590	1 in 29,000	1 in 120,000
		Latino/Admixed American	98	1 in 220	1 in 11,000	1 in 44,000
		Ashkenazi Jewish	99	1 in 72	1 in 7,100	1 in 28,000
		East Asian	99	1 in 1,124	1 in 110,000	1 in 440,000
		Finnish	99	1 in 511	1 in 51,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 180	1 in 18,000	1 in 72,000
		South Asian	99	1 in 411	1 in 41,000	1 in 160,000
		Other (population not assigned)	99	1 in 139	1 in 14,000	1 in 56,000
GCDH	Glutaric acidemia type I	African/African American	99	1 in 89	1 in 8,800	1 in 35,000
		Latino/Admixed American	99	1 in 234	1 in 23,000	1 in 92,000
		Ashkenazi Jewish	99	1 in 1,680	1 in 170,000	1 in 680,000
		East Asian	99	1 in 212	1 in 21,000	1 in 84,000
		Finnish	99	1 in 507	1 in 51,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 219	1 in 22,000	1 in 88,000
		South Asian	98	1 in 172	1 in 8,600	1 in 34,000
		Other (population not assigned)	99	1 in 254	1 in 25,000	1 in 100,000
GFPT1	Congenital myasthenic syndrome	African/African American	99	1 in 1,012	1 in 100,000	1 in 400,000
		Latino/Admixed American	99	1 in 293	1 in 29,000	1 in 120,000
		Ashkenazi Jewish	99	1 in 2,514	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 1,729	1 in 170,000	1 in 680,000
		Finnish	99	1 in 974	1 in 97,000	1 in 390,000
		Non-Finnish European/Caucasian	99	1 in 192	1 in 19,000	1 in 76,000
		South Asian	98	1 in 7,652	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	1 in 231	1 in 23,000	1 in 92,000
GJB2	Deafness and hearing loss, nonsyndromic	African/African American	99	1 in 113	1 in 11,000	1 in 44,000
		Latino/Admixed American	95	1 in 37	1 in 720	1 in 2,900
		Ashkenazi Jewish	99	1 in 57	1 in 5,600	1 in 22,000
		East Asian	97	1 in 58	1 in 1,900	1 in 7,600
		Finnish	97	1 in 58	1 in 1,900	1 in 7,600
		Non-Finnish European/Caucasian	98	1 in 38	1 in 1,900	1 in 7,600
		South Asian	99	1 in 58	1 in 5,700	1 in 23,000
		Other (population not assigned)	98	1 in 47	1 in 2,300	1 in 9,200
GJB6	Deafness and hearing loss, nonsyndromic	African/African American	97	1 in 1,626	1 in 54,000	1 in 220,000
		Latino/Admixed American	71	1 in 2,161	1 in 7,400	1 in 30,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Ashkenazi Jewish	99	1 in 5,032	1 in 500,000	1 in 2,000,000
		East Asian	98	1 in 214	1 in 11,000	1 in 44,000
		Finnish	97	1 in 5,411	1 in 180,000	1 in 720,000
		Non-Finnish European/Caucasian	94	1 in 421	1 in 7,000	1 in 28,000
		South Asian	99	1 in 1,531	1 in 150,000	1 in 600,000
		Other (population not assigned)	97	1 in 256	1 in 8,500	1 in 34,000
GLA	Fabry disease	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
GLB1	GM1 gangliosidosis and mucopolysaccharidosis type IVB	African/African American	98	1 in 272	1 in 14,000	1 in 56,000
		Latino/Admixed American	99	1 in 499	1 in 50,000	1 in 200,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 489	1 in 49,000	1 in 200,000
		Finnish	98	1 in 251	1 in 13,000	1 in 52,000
		Non-Finnish European/Caucasian	99	1 in 343	1 in 34,000	1 in 140,000
		South Asian	99	1 in 239	1 in 24,000	1 in 96,000
		Other (population not assigned)	99	1 in 402	1 in 40,000	1 in 160,000
GLDC	Glycine encephalopathy	African/African American	99	1 in 1,141	1 in 110,000	1 in 440,000
		Latino/Admixed American	99	1 in 584	1 in 58,000	1 in 230,000
		Ashkenazi Jewish	98	1 in 2,518	1 in 130,000	1 in 520,000
		East Asian	99	1 in 460	1 in 46,000	1 in 180,000
		Finnish	99	1 in 118	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	99	1 in 388	1 in 39,000	1 in 160,000
		South Asian	98	1 in 368	1 in 18,000	1 in 72,000
		Other (population not assigned)	99	1 in 457	1 in 46,000	1 in 180,000
GNE	Inclusion body myopathy 2	African/African American	98	1 in 249	1 in 12,000	1 in 48,000
		Latino/Admixed American	99	1 in 793	1 in 79,000	1 in 320,000
		Ashkenazi Jewish	99	1 in 4,924	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 101	1 in 10,000	1 in 40,000
		Finnish	98	1 in 1,387	1 in 69,000	1 in 280,000
		Non-Finnish European/Caucasian	99	1 in 368	1 in 37,000	1 in 150,000
		South Asian	99	1 in 36	1 in 3,500	1 in 14,000
		Other (population not assigned)	99	1 in 252	1 in 25,000	1 in 100,000
GNPAT	Rhizomelic chondrodysplasia punctata	African/African American	99	1 in 2,709	1 in 270,000	1 in 1,100,000
		Latino/Admixed American	99	1 in 5,707	1 in 570,000	1 in 2,300,000
		Ashkenazi Jewish	97	1 in 5,041	1 in 170,000	1 in 680,000
		East Asian	98	1 in 4,450	1 in 220,000	1 in 880,000
		Finnish	98	1 in 5,478	1 in 270,000	1 in 1,100,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Non-Finnish European/Caucasian	99	1 in 3,136	1 in 310,000	1 in 1,200,000
		South Asian	98	1 in 5,102	1 in 260,000	1 in 1,000,000
		Other (population not assigned)	97	N/A	N/A	N/A
GNPTAB	Mucolipidosis type II and III	African/African American	97	1 in 114	1 in 3,800	1 in 15,000
		Latino/Admixed American	97	1 in 123	1 in 4,100	1 in 16,000
		Ashkenazi Jewish	97	1 in 281	1 in 9,300	1 in 37,000
		East Asian	98	1 in 265	1 in 13,000	1 in 52,000
		Finnish	97	1 in 99	1 in 3,300	1 in 13,000
		Non-Finnish European/Caucasian	97	1 in 163	1 in 5,400	1 in 22,000
		South Asian	97	1 in 114	1 in 3,800	1 in 15,000
		Other (population not assigned)	97	1 in 139	1 in 4,600	1 in 18,000
GNS	Mucopolysaccharidosis type III	African/African American	98	1 in 3,825	1 in 190,000	1 in 760,000
		Latino/Admixed American	99	1 in 793	1 in 79,000	1 in 320,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 4,588	1 in 460,000	1 in 1,800,000
		Finnish	97	1 in 10,825	1 in 360,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 1,961	1 in 200,000	1 in 800,000
		South Asian	98	1 in 4,145	1 in 210,000	1 in 840,000
		Other (population not assigned)	99	1 in 3,066	1 in 310,000	1 in 1,200,000
GRHPR	Primary hyperoxaluria	African/African American	99	1 in 1,836	1 in 180,000	1 in 720,000
		Latino/Admixed American	98	1 in 557	1 in 28,000	1 in 110,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 288	1 in 9,600	1 in 38,000
		Finnish	99	1 in 636	1 in 64,000	1 in 260,000
		Non-Finnish European/Caucasian	98	1 in 353	1 in 18,000	1 in 72,000
		South Asian	99	1 in 222	1 in 22,000	1 in 88,000
		Other (population not assigned)	99	1 in 573	1 in 57,000	1 in 230,000
GRIP1	Fraser syndrome	African/African American	98	1 in 669	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 187	1 in 19,000	1 in 76,000
		Ashkenazi Jewish	99	1 in 82	1 in 8,100	1 in 32,000
		East Asian	99	1 in 1,032	1 in 100,000	1 in 400,000
		Finnish	97	1 in 901	1 in 30,000	1 in 120,000
		Non-Finnish European/Caucasian	99	1 in 333	1 in 33,000	1 in 130,000
		South Asian	99	1 in 314	1 in 31,000	1 in 120,000
		Other (population not assigned)	99	1 in 137	1 in 14,000	1 in 56,000
GSS	Glutathione synthetase deficiency	African/African American	99	1 in 49	1 in 4,800	1 in 19,000
		Latino/Admixed American	99	1 in 618	1 in 62,000	1 in 250,000
		Ashkenazi Jewish	97	1 in 112	1 in 3,700	1 in 15,000
		East Asian	99	1 in 297	1 in 30,000	1 in 120,000
		Finnish	99	1 in 3,680	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	99	1 in 992	1 in 99,000	1 in 400,000
		South Asian	98	1 in 1,703	1 in 85,000	1 in 340,000
		Other (population not assigned)	98	1 in 438	1 in 22,000	1 in 88,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
GUSB	Mucopolysaccharidosis type VII	African/African American	99	1 in 488	1 in 49,000	1 in 200,000
		Latino/Admixed American	99	1 in 685	1 in 68,000	1 in 270,000
		Ashkenazi Jewish	99	1 in 741	1 in 74,000	1 in 300,000
		East Asian	99	1 in 1,760	1 in 180,000	1 in 720,000
		Finnish	99	1 in 1,177	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	99	1 in 552	1 in 55,000	1 in 220,000
		South Asian	99	1 in 914	1 in 91,000	1 in 360,000
		Other (population not assigned)	99	1 in 661	1 in 66,000	1 in 260,000
HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	African/African American	99	1 in 463	1 in 46,000	1 in 180,000
		Latino/Admixed American	99	1 in 393	1 in 39,000	1 in 160,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	1 in 537	1 in 18,000	1 in 72,000
		Finnish	99	1 in 117	1 in 12,000	1 in 48,000
		Non-Finnish European/Caucasian	99	1 in 212	1 in 21,000	1 in 84,000
		South Asian	99	1 in 698	1 in 70,000	1 in 280,000
		Other (population not assigned)	99	1 in 196	1 in 20,000	1 in 80,000
HADHB	Trifunctional protein deficiency	African/African American	99	1 in 1,095	1 in 110,000	1 in 440,000
		Latino/Admixed American	99	1 in 827	1 in 83,000	1 in 330,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 742	1 in 74,000	1 in 300,000
		Finnish	97	1 in 2,165	1 in 72,000	1 in 290,000
		Non-Finnish European/Caucasian	99	1 in 1,379	1 in 140,000	1 in 560,000
		South Asian	99	1 in 1,033	1 in 100,000	1 in 400,000
		Other (population not assigned)	98	1 in 1,448	1 in 72,000	1 in 290,000
HAMP	Juvenile hereditary hemochromatosis	African/African American	98	1 in 4,059	1 in 200,000	1 in 800,000
		Latino/Admixed American	98	N/A	N/A	N/A
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 1,150	1 in 110,000	1 in 440,000
		Finnish	98	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 8,124	1 in 810,000	1 in 3,200,000
		South Asian	99	1 in 7,653	1 in 770,000	1 in 3,100,000
		Other (population not assigned)	98	N/A	N/A	N/A
HAX1	Severe congenital neutropenia, autosomal recessive	African/African American	99	1 in 569	1 in 57,000	1 in 230,000
		Latino/Admixed American	99	1 in 1,922	1 in 190,000	1 in 760,000
		Ashkenazi Jewish	97	1 in 837	1 in 28,000	1 in 110,000
		East Asian	99	1 in 563	1 in 56,000	1 in 220,000
		Finnish	97	1 in 472	1 in 16,000	1 in 64,000
		Non-Finnish European/Caucasian	97	1 in 769	1 in 26,000	1 in 100,000
		South Asian	99	1 in 219	1 in 22,000	1 in 88,000
		Other (population not assigned)	99	1 in 599	1 in 60,000	1 in 240,000
HBA1/HBA2	Alpha-thalassemia	African	90	1 in 3	N/A	N/A
		American	90	1 in 21	N/A	N/A
		Eastern Mediterranean	90	1 in 5	N/A	N/A

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		European	90	1 in 44	N/A	N/A
		Southeast Asian	90	1 in 2	N/A	N/A
		Western Pacific	90	1 in 10	N/A	N/A
HBB	Beta-hemoglobinopathies, includes sickle cell disease and beta-thalassemias	African/African American	99	1 in 9	1 in 800	1 in 3,200
		Latino/Admixed American	99	1 in 164	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	99	1 in 4,922	1 in 490,000	1 in 2,000,000
		East Asian	98	1 in 117	1 in 5,800	1 in 23,000
		Finnish	99	1 in 140	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 240	1 in 24,000	1 in 96,000
		South Asian	99	1 in 27	1 in 2,600	1 in 10,000
		Other (population not assigned)	99	1 in 77	1 in 7,600	1 in 30,000
HCFC1	Methylmalonic acidemia with homocystinuria	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	97	N/A	N/A	N/A
		South Asian	97	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
HELLS	Immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome	African/African American	97	1 in 1,081	1 in 36,000	1 in 140,000
		Latino/Admixed American	97	1 in 481	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	97	1 in 1,222	1 in 41,000	1 in 160,000
		East Asian	99	1 in 413	1 in 41,000	1 in 160,000
		Finnish	98	1 in 1,793	1 in 90,000	1 in 360,000
		Non-Finnish European/Caucasian	98	1 in 1,155	1 in 58,000	1 in 230,000
		South Asian	98	1 in 674	1 in 34,000	1 in 140,000
		Other (population not assigned)	98	1 in 624	1 in 31,000	1 in 120,000
HEXA	Tay-Sachs disease	African/African American	98	1 in 132	1 in 6,600	1 in 26,000
		Latino/Admixed American	99	1 in 255	1 in 25,000	1 in 100,000
		Ashkenazi Jewish	97	1 in 30	1 in 970	1 in 3,900
		East Asian	98	1 in 468	1 in 23,000	1 in 92,000
		Finnish	98	1 in 366	1 in 18,000	1 in 72,000
		Non-Finnish European/Caucasian	99	1 in 91	1 in 9,000	1 in 36,000
		South Asian	99	1 in 379	1 in 38,000	1 in 150,000
		Other (population not assigned)	98	1 in 170	1 in 8,500	1 in 34,000
HEXB	Sandhoff disease	African/African American	98	1 in 883	1 in 44,000	1 in 180,000
		Latino/Admixed American	98	1 in 330	1 in 16,000	1 in 64,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	99	1 in 394	1 in 39,000	1 in 160,000
		Finnish	99	1 in 3,546	1 in 350,000	1 in 1,400,000
		Non-Finnish European/Caucasian	99	1 in 267	1 in 27,000	1 in 110,000
		South Asian	99	1 in 529	1 in 53,000	1 in 210,000
		Other (population not assigned)	98	1 in 490	1 in 24,000	1 in 96,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
HGSNAT	Mucopolysaccharidosis type III	African/African American	99	1 in 696	1 in 70,000	1 in 280,000
		Latino/Admixed American	99	1 in 730	1 in 73,000	1 in 290,000
		Ashkenazi Jewish	97	1 in 1,480	1 in 49,000	1 in 200,000
		East Asian	99	1 in 858	1 in 86,000	1 in 340,000
		Finnish	98	1 in 775	1 in 39,000	1 in 160,000
		Non-Finnish European/Caucasian	99	1 in 770	1 in 77,000	1 in 310,000
		South Asian	99	1 in 976	1 in 98,000	1 in 390,000
		Other (population not assigned)	99	1 in 924	1 in 92,000	1 in 370,000
HINT1	Axonal neuropathy with neuromyotonia, autosomal recessive	African/African American	99	1 in 1,824	1 in 180,000	1 in 720,000
		Latino/Admixed American	99	1 in 2,188	1 in 220,000	1 in 880,000
		Ashkenazi Jewish	99	1 in 179	1 in 18,000	1 in 72,000
		East Asian	99	1 in 999	1 in 100,000	1 in 400,000
		Finnish	99	1 in 578	1 in 58,000	1 in 230,000
		Non-Finnish European/Caucasian	99	1 in 766	1 in 77,000	1 in 310,000
		South Asian	99	1 in 3,178	1 in 320,000	1 in 1,300,000
		Other (population not assigned)	99	1 in 2,739	1 in 270,000	1 in 1,100,000
HJV	Juvenile hereditary hemochromatosis	African/African American	97	N/A	N/A	N/A
		Latino/Admixed American	97	1 in 3,456	1 in 120,000	1 in 480,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 2,225	1 in 220,000	1 in 880,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 858	1 in 86,000	1 in 340,000
		South Asian	97	1 in 766	1 in 26,000	1 in 100,000
		Other (population not assigned)	99	1 in 1,449	1 in 140,000	1 in 560,000
HLCS	Holocarboxylase synthetase deficiency	African/African American	97	1 in 985	1 in 33,000	1 in 130,000
		Latino/Admixed American	99	1 in 1,073	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	97	1 in 5,041	1 in 170,000	1 in 680,000
		East Asian	99	1 in 366	1 in 37,000	1 in 150,000
		Finnish	99	1 in 3,717	1 in 370,000	1 in 1,500,000
		Non-Finnish European/Caucasian	99	1 in 920	1 in 92,000	1 in 370,000
		South Asian	98	1 in 958	1 in 48,000	1 in 190,000
		Other (population not assigned)	98	1 in 724	1 in 36,000	1 in 140,000
HMGCL	HMG-CoA lyase deficiency	African/African American	99	1 in 440	1 in 44,000	1 in 180,000
		Latino/Admixed American	99	1 in 130	1 in 13,000	1 in 52,000
		Ashkenazi Jewish	98	1 in 2,244	1 in 110,000	1 in 440,000
		East Asian	99	1 in 2,226	1 in 220,000	1 in 880,000
		Finnish	99	1 in 490	1 in 49,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 102	1 in 10,000	1 in 40,000
		South Asian	98	1 in 197	1 in 9,800	1 in 39,000
		Other (population not assigned)	99	1 in 136	1 in 14,000	1 in 56,000
HOGA1	Primary hyperoxaluria	African/African American	99	1 in 287	1 in 29,000	1 in 120,000
		Latino/Admixed American	99	1 in 267	1 in 27,000	1 in 110,000
		Ashkenazi Jewish	98	1 in 37	1 in 1,800	1 in 7,200

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		East Asian	99	1 in 182	1 in 18,000	1 in 72,000
		Finnish	99	1 in 507	1 in 51,000	1 in 200,000
		Non-Finnish European/Caucasian	99	1 in 167	1 in 17,000	1 in 68,000
		South Asian	99	1 in 265	1 in 26,000	1 in 100,000
		Other (population not assigned)	99	1 in 145	1 in 14,000	1 in 56,000
HPD	Tyrosinemia type III	African/African American	98	1 in 4,064	1 in 200,000	1 in 800,000
		Latino/Admixed American	98	1 in 5,764	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	99	1 in 1,680	1 in 170,000	1 in 680,000
		East Asian	97	1 in 9,196	1 in 310,000	1 in 1,200,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 2,453	1 in 250,000	1 in 1,000,000
		South Asian	99	1 in 1,026	1 in 100,000	1 in 400,000
		Other (population not assigned)	97	N/A	N/A	N/A
HPS1	Hermansky-Pudlak syndrome	African/African American	98	1 in 2,695	1 in 130,000	1 in 520,000
		Latino/Admixed American	98	1 in 1,057	1 in 53,000	1 in 210,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,279	1 in 110,000	1 in 440,000
		Finnish	99	1 in 10,813	1 in 1,100,000	1 in 4,400,000
		Non-Finnish European/Caucasian	97	1 in 1,250	1 in 42,000	1 in 170,000
		South Asian	99	1 in 1,085	1 in 110,000	1 in 440,000
		Other (population not assigned)	97	N/A	N/A	N/A
HPS3	Hermansky-Pudlak syndrome	African/African American	98	1 in 541	1 in 27,000	1 in 110,000
		Latino/Admixed American	99	1 in 1,150	1 in 110,000	1 in 440,000
		Ashkenazi Jewish	99	1 in 242	1 in 24,000	1 in 96,000
		East Asian	99	1 in 289	1 in 29,000	1 in 120,000
		Finnish	98	1 in 524	1 in 26,000	1 in 100,000
		Non-Finnish European/Caucasian	99	1 in 590	1 in 59,000	1 in 240,000
		South Asian	99	1 in 322	1 in 32,000	1 in 130,000
		Other (population not assigned)	99	1 in 703	1 in 70,000	1 in 280,000
HPS4	Hermansky-Pudlak syndrome	African/African American	98	1 in 1,402	1 in 70,000	1 in 280,000
		Latino/Admixed American	99	1 in 384	1 in 38,000	1 in 150,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,289	1 in 110,000	1 in 440,000
		Finnish	98	1 in 5,412	1 in 270,000	1 in 1,100,000
		Non-Finnish European/Caucasian	99	1 in 703	1 in 70,000	1 in 280,000
		South Asian	98	1 in 2,003	1 in 100,000	1 in 400,000
		Other (population not assigned)	97	1 in 745	1 in 25,000	1 in 100,000
HPS5	Hermansky-Pudlak syndrome	African/African American	97	1 in 69	1 in 2,300	1 in 9,200
		Latino/Admixed American	99	1 in 202	1 in 20,000	1 in 80,000
		Ashkenazi Jewish	97	1 in 275	1 in 9,100	1 in 36,000
		East Asian	98	1 in 471	1 in 24,000	1 in 96,000
		Finnish	99	1 in 145	1 in 14,000	1 in 56,000
		Non-Finnish European/Caucasian	99	1 in 89	1 in 8,800	1 in 35,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		South Asian	99	1 in 201	1 in 20,000	1 in 80,000
		Other (population not assigned)	99	1 in 134	1 in 13,000	1 in 52,000
HPS6	Hermansky-Pudlak syndrome	African/African American	98	1 in 1,121	1 in 56,000	1 in 220,000
		Latino/Admixed American	97	1 in 1,310	1 in 44,000	1 in 180,000
		Ashkenazi Jewish	97	1 in 4,990	1 in 170,000	1 in 680,000
		East Asian	98	1 in 1,090	1 in 54,000	1 in 220,000
		Finnish	99	1 in 1,352	1 in 140,000	1 in 560,000
		Non-Finnish European/Caucasian	98	1 in 745	1 in 37,000	1 in 150,000
		South Asian	98	1 in 1,767	1 in 88,000	1 in 350,000
		Other (population not assigned)	98	1 in 1,519	1 in 76,000	1 in 300,000
HSD3B2	Congenital adrenal hyperplasia	African/African American	97	1 in 1,151	1 in 38,000	1 in 150,000
		Latino/Admixed American	99	1 in 2,853	1 in 290,000	1 in 1,200,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 2,258	1 in 230,000	1 in 920,000
		Finnish	99	1 in 5,495	1 in 550,000	1 in 2,200,000
		Non-Finnish European/Caucasian	99	1 in 1,712	1 in 170,000	1 in 680,000
		South Asian	98	1 in 1,183	1 in 59,000	1 in 240,000
		Other (population not assigned)	97	N/A	N/A	N/A
HSD17B4	D-bifunctional protein deficiency	African/African American	98	1 in 326	1 in 16,000	1 in 64,000
		Latino/Admixed American	99	1 in 638	1 in 64,000	1 in 260,000
		Ashkenazi Jewish	99	1 in 2,490	1 in 250,000	1 in 1,000,000
		East Asian	99	1 in 449	1 in 45,000	1 in 180,000
		Finnish	99	1 in 293	1 in 29,000	1 in 120,000
		Non-Finnish European/Caucasian	99	1 in 424	1 in 42,000	1 in 170,000
		South Asian	99	1 in 122	1 in 12,000	1 in 48,000
		Other (population not assigned)	99	1 in 2,742	1 in 270,000	1 in 1,100,000
HSD17B10	HSD10 disease	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	99	N/A	N/A	N/A
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
HYAL1	Mucopolysaccharidosis type IX	African/African American	97	1 in 2,692	1 in 90,000	1 in 360,000
		Latino/Admixed American	98	1 in 1,708	1 in 85,000	1 in 340,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	98	1 in 2,296	1 in 110,000	1 in 440,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	98	1 in 2,811	1 in 140,000	1 in 560,000
		South Asian	98	1 in 1,691	1 in 85,000	1 in 340,000
		Other (population not assigned)	97	N/A	N/A	N/A
HYLS1	Hydrolethalus syndrome	African/African American	98	1 in 670	1 in 33,000	1 in 130,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

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		Latino/Admixed American	99	1 in 592	1 in 59,000	1 in 240,000
		Ashkenazi Jewish	99	1 in 4,925	1 in 490,000	1 in 2,000,000
		East Asian	99	1 in 1,150	1 in 110,000	1 in 440,000
		Finnish	99	1 in 54	1 in 5,300	1 in 21,000
		Non-Finnish European/Caucasian	99	1 in 443	1 in 44,000	1 in 180,000
		South Asian	98	1 in 487	1 in 24,000	1 in 96,000
		Other (population not assigned)	99	1 in 348	1 in 35,000	1 in 140,000
IDS	Mucopolysaccharidosis type II	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	97	N/A	N/A	N/A
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	97	N/A	N/A	N/A
IDUA	Mucopolysaccharidosis type I	African/African American	99	1 in 515	1 in 51,000	1 in 200,000
		Latino/Admixed American	98	1 in 119	1 in 5,900	1 in 24,000
		Ashkenazi Jewish	97	1 in 224	1 in 7,400	1 in 30,000
		East Asian	99	1 in 55	1 in 5,400	1 in 22,000
		Finnish	98	1 in 88	1 in 4,400	1 in 18,000
		Non-Finnish European/Caucasian	99	1 in 111	1 in 11,000	1 in 44,000
		South Asian	98	1 in 214	1 in 11,000	1 in 44,000
		Other (population not assigned)	99	1 in 165	1 in 16,000	1 in 64,000
IFT140	Retinitis pigmentosa	African/African American	98	1 in 315	1 in 16,000	1 in 64,000
		Latino/Admixed American	99	1 in 382	1 in 38,000	1 in 150,000
		Ashkenazi Jewish	99	1 in 180	1 in 18,000	1 in 72,000
		East Asian	99	1 in 269	1 in 27,000	1 in 110,000
		Finnish	98	1 in 1,067	1 in 53,000	1 in 210,000
		Non-Finnish European/Caucasian	99	1 in 381	1 in 38,000	1 in 150,000
		South Asian	99	1 in 247	1 in 25,000	1 in 100,000
		Other (population not assigned)	99	1 in 474	1 in 47,000	1 in 190,000
IKBKB	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 405	1 in 40,000	1 in 160,000
		Latino/Admixed American	99	1 in 259	1 in 26,000	1 in 100,000
		Ashkenazi Jewish	99	1 in 260	1 in 26,000	1 in 100,000
		East Asian	97	1 in 470	1 in 16,000	1 in 64,000
		Finnish	99	1 in 962	1 in 96,000	1 in 380,000
		Non-Finnish European/Caucasian	99	1 in 324	1 in 32,000	1 in 130,000
		South Asian	99	1 in 427	1 in 43,000	1 in 170,000
		Other (population not assigned)	99	1 in 187	1 in 19,000	1 in 76,000
IL2RA	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 674	1 in 67,000	1 in 270,000
		Latino/Admixed American	99	1 in 5,765	1 in 580,000	1 in 2,300,000
		Ashkenazi Jewish	99	N/A	N/A	N/A
		East Asian	99	1 in 2,300	1 in 230,000	1 in 920,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 11,327	1 in 1,100,000	1 in 4,400,000
		South Asian	99	1 in 3,827	1 in 380,000	1 in 1,500,000
		Other (population not assigned)	99	N/A	N/A	N/A
IL2RG	Severe combined Immunodeficiency (SCID), X-linked	African/African American	99	N/A	N/A	N/A
		Latino/Admixed American	97	N/A	N/A	N/A
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	N/A	N/A	N/A
		Finnish	99	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	N/A	N/A	N/A
		South Asian	99	N/A	N/A	N/A
		Other (population not assigned)	99	N/A	N/A	N/A
IL7R	Severe combined immunodeficiency (SCID)	African/African American	99	1 in 471	1 in 47,000	1 in 190,000
		Latino/Admixed American	99	1 in 426	1 in 43,000	1 in 170,000
		Ashkenazi Jewish	98	N/A	N/A	N/A
		East Asian	98	1 in 1,839	1 in 92,000	1 in 370,000
		Finnish	99	1 in 4,073	1 in 410,000	1 in 1,600,000
		Non-Finnish European/Caucasian	99	1 in 522	1 in 52,000	1 in 210,000
		South Asian	98	1 in 1,682	1 in 84,000	1 in 340,000
		Other (population not assigned)	99	1 in 764	1 in 76,000	1 in 300,000
INPP5E	Joubert syndrome and related disorders, including Meckel-Gruber syndrome	African/African American	98	1 in 397	1 in 20,000	1 in 80,000
		Latino/Admixed American	99	1 in 1,685	1 in 170,000	1 in 680,000
		Ashkenazi Jewish	97	N/A	N/A	N/A
		East Asian	99	1 in 1,454	1 in 150,000	1 in 600,000
		Finnish	97	1 in 735	1 in 24,000	1 in 96,000
		Non-Finnish European/Caucasian	97	1 in 156	1 in 5,200	1 in 21,000
		South Asian	98	1 in 1,600	1 in 80,000	1 in 320,000
		Other (population not assigned)	99	1 in 1,933	1 in 190,000	1 in 760,000
ITPA	Early infantile epileptic encephalopathy	African/African American	98	1 in 3,918	1 in 200,000	1 in 800,000
		Latino/Admixed American	99	1 in 5,707	1 in 570,000	1 in 2,300,000
		Ashkenazi Jewish	97	1 in 4,918	1 in 160,000	1 in 640,000
		East Asian	98	1 in 1,838	1 in 92,000	1 in 370,000
		Finnish	99	1 in 1,195	1 in 120,000	1 in 480,000
		Non-Finnish European/Caucasian	98	1 in 2,093	1 in 100,000	1 in 400,000
		South Asian	99	1 in 7,654	1 in 770,000	1 in 3,100,000
		Other (population not assigned)	99	1 in 3,068	1 in 310,000	1 in 1,200,000
IVD	Isovaleric acidemia	African/African American	98	1 in 461	1 in 23,000	1 in 92,000
		Latino/Admixed American	99	1 in 360	1 in 36,000	1 in 140,000
		Ashkenazi Jewish	99	1 in 420	1 in 42,000	1 in 170,000
		East Asian	99	1 in 303	1 in 30,000	1 in 120,000
		Finnish	98	1 in 1,099	1 in 55,000	1 in 220,000
		Non-Finnish European/Caucasian	99	1 in 232	1 in 23,000	1 in 92,000
		South Asian	99	1 in 507	1 in 51,000	1 in 200,000

Detection Rates, Pre-Test Carrier Risks, and Post-Test Residual Risks (Sorted by Gene)

The detection rates and risks set forth below were derived using publicly available information, including gnomAD and ClinVar, and the rates were extrapolated using appropriate scientific methodologies. As additional clinical evidence is available, the data in these charts may be updated from time to time. This data is provided for general informational purposes only and is not intended as a substitute for professional medical judgment or medical advice.

For unknown or mixed ethnicity, consider using the ethnic background with the most conservative risk estimates.

N/A= Not Available

Gene	Disorder	Ethnicity (Source: https://gnomad.broadinstitute.org/ except CYP21A2 and HBA1/HBA2)	Detection Rate (%) (Derived from gnomAD and ClinVar except CYP21A2, GBA, and HBA1/HBA2)	Individual Carrier Risk Before Testing (Derived from gnomAD and ClinVar except CYP21A2 and HBA1/HBA2)	Individual Residual Risk After Negative Result	Risk of Affected Fetus When One Partner has a Carrier Result and One Partner has a Negative Result
		Other (population not assigned)	99	1 in 207	1 in 21,000	1 in 84,000
JAK3	Severe combined immunodeficiency (SCID)	African/African American	98	1 in 803	1 in 40,000	1 in 160,000
		Latino/Admixed American	99	1 in 174	1 in 17,000	1 in 68,000
		Ashkenazi Jewish	99	1 in 85	1 in 8,400	1 in 34,000
		East Asian	99	1 in 1,661	1 in 170,000	1 in 680,000
		Finnish	97	N/A	N/A	N/A
		Non-Finnish European/Caucasian	99	1 in 475	1 in 47,000	1 in 190,000
		South Asian	97	1 in 686	1 in 23,000	1 in 92,000
		Other (population not assigned)	99	1 in 196	1 in 20,000	1 in 80,000